ALX3 gene

ALX homeobox 3

Normal Function

The *ALX3* gene provides instructions for making a protein that is a member of the homeobox protein family. Homeobox proteins direct the formation of body structures during early embryonic development. The ALX3 protein is necessary for normal development of the head and face, particularly the formation of the nose, which begins around the fourth week of development. The ALX3 protein is a transcription factor, which means that it attaches (binds) to DNA and controls the activity of certain genes. Specifically, the protein controls the activity of genes that regulate cell growth and division (proliferation) and movement (migration), ensuring that cells grow and stop growing at specific times and that they are positioned correctly during development.

Health Conditions Related to Genetic Changes

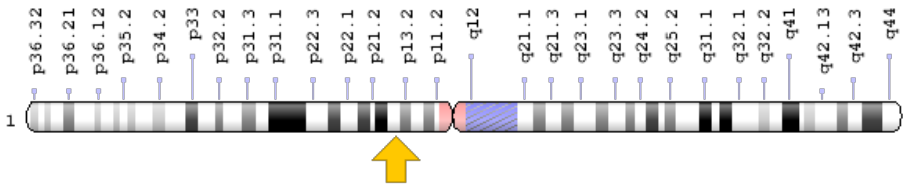
frontonasal dysplasia

At least seven mutations in the *ALX3* gene have been found to cause frontonasal dysplasia. *ALX3* gene mutations cause a form of the disorder called frontonasal dysplasia type 1, which particularly affects the development of the nose and surrounding tissues. *ALX3* gene mutations that cause this condition severely reduce or eliminate the function of the ALX3 protein. As a result, the protein cannot bind to DNA and regulate gene function, which leads to poorly controlled cell proliferation and migration during development. This abnormal cell growth and movement impairs development of structures in the middle of the face, particularly the nose, leading to openings (clefts) in the nose. This abnormal development can also interfere with the proper formation of the skull, leading to the skull malformations typical of frontonasal dysplasia type 1.

Chromosomal Location

Cytogenetic Location: 1p13.3, which is the short (p) arm of chromosome 1 at position 13.3

Molecular Location: base pairs 110,060,375 to 110,070,700 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- aristaless-like homeobox 3
- FND
- FND1
- frontonasal dysplasia
- homeobox protein aristaless-like 3
- proline-rich transcription factor ALX3

Additional Information & Resources

Educational Resources

- Jasper's Basic Mechanisms of the Epilepsies (fourth edition, 2012): Molecular Biology of ARX
  https://www.ncbi.nlm.nih.gov/books/NBK98176/#marsh.s3

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALX3%5BTIAB%5D%29+OR+%28aristaless-like+homeobox+3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- ARISTALESS-LIKE HOMEOBOX 3
  http://omim.org/entry/606014

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALX3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALX3%5Bgene%5D
- HGNC Gene Family: PRD class homeoboxes and pseudogenes
  http://www.genenames.org/cgi-bin/genefamilies/set/521
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=449
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/257
- UniProt
  http://www.uniprot.org/uniprot/O95076

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